

Suggested Follow-up for Congenital Adrenal Hyperplasia (CAH)  
Elevated 17-OH Progesterone (17-OHP)

Possible Causes: Congenital adrenal hyperplasia (CAH) occurs when a metabolic block produces varying degrees of insufficiency of corticosteroids (cortisone and aldosterone) and an excess of male sex hormones. Elevated 17-OHP is also commonly found in premature and/or stressed infants.

Next Steps if Abnormal: **Potential medical emergency when 17-OHP is significantly elevated.** See infant as soon as possible to ascertain health status. Repeat 17-OHP screening on filter paper and send to the DHEC laboratory. Consider measurement of serum electrolytes and consult pediatric endocrinologist for further instructions if 17-OHP is over 48 ng/mL in infants  $\geq$  2500 g or over 130 ng/mL in infants  $<$  2500 g.

Neonatal Presentation: Salt-wasting CAH: Acute crisis with failure to thrive, dehydration and shock. All forms of CAH: females may have ambiguous genitalia.

Emergency Treatment: Provide replacement hormones and correct electrolyte imbalances as indicated.

Standard Treatment: Provide replacement hormones. Increased dosages of medications are usually required in times of stress, trauma, illness or surgery.

Advice for Family: Provide basic information about congenital adrenal hyperplasia. The handout, *When Baby Needs a Second Test for Congenital Adrenal Hyperplasia*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

Internet Resources:

<http://www.genetests.org/query?dz=cah>

<http://www.every1cares.org/>

<http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm>